Congenital Heart Defects

Objectives
- Review Normal Cardiac Anatomy
- Discuss Basics of Fetal Cardiac Development
- Discuss Common Congenital Heart Defects
- Review the AHA Guidelines
- Treatment Objectives
- Ask a Ninja
Normal Cardiac Anatomy Review

- The human heart is a strong, hard working pump about the size of a person’s fist.

- The heart is divided into four chambers:
  - The two upper chambers are the atria.
  - The two lower chambers are the ventricles.
  - The atria and ventricles are divided by tissue, known as “septa.”
Normal Cardiac Anatomy Review

- There are four valves in the heart that keep blood flowing in one direction only.
- Congenital defects may involve a valve, a chamber, the septum, an artery or the flow of blood.
Normal Cardiac Anatomy Review

- Four Chamber Heart
  - Right Sided Circulation
    - Deoxygenated blood flow through the lungs and pulmonary vasculature
    - Superior and Inferior Vena Cava to Right Atrium
    - Through the Tricuspid Valve to the Right Ventricle
    - Through the Pulmonary Valve to the lungs
  - Left Sided Circulation
    - Oxygenated blood flow to the body
    - Pulmonary veins to the Left Atrium
    - Through the Mitral Valve to the Left Ventricle
    - Through the Aortic Valve to the body
How do we get from two cells joining together to a huge, complex organism carrying out complex functions? 

Embryology!!!

- An entire field of science dedicated to studying the development of the human embryo
- Looks at many, complex biological markers and intracellular signals that make cells differentiate and specialize
- Signals have to be exactly right, at exactly the right time to make sure the development happens in the correct order
- Researchers have spent lifetimes looking at this
- Embryology takes an entire year of coursework in medical school
- Development of the heart alone could take a week to just scratch the surface
Embryology

- We’re going to cover it all in about 20 minutes.
- So the question – as always – is...
- What do we need to know?
  - First of all, the human body truly is a miracle
  - I am not an Embryologist
    - There is a huge amount of this that I struggle to understand myself
    - Some of the mechanics are still not known, thought to be related to different gene expressions, beating of cilia, etc
  - The heart is one example of the multitude of how a cascade of complex biochemical reactions can turn two sets of tubes into a highly specialized system, designed to exchange gases and carry out functions of life
So what do we need to know?

- Heart development in the embryo begins with two separate tubes that join together
- Venous end, and arterial end

http://en.wikipedia.org/wiki/Heart_development
So what do we need to know?
- The heart begins beating approximately 21 days after conception
- Then undergoes rightward looping
Embryology

- So what do we need to know?
  - The tube then subdivides the ventricles from the atria, and the right from the left
  - The truncus arteriosus splits to form the aorta and the pulmonary veins
  - This is the point during development during which many of the errors can occur
  - We will talk about these in a little bit
    - For now, just keep these images in the back of your mind…
“Mr. Osborne, may I be excused? My brain is full.”
Consult The Interwebs

If your confused like me, there are some cool websites you can visit for more information

- [http://pie.med.utoronto.ca/htbg/HTBG_content/HTBG_heartEmbryologyApp.html](http://pie.med.utoronto.ca/htbg/HTBG_content/HTBG_heartEmbryologyApp.html)
- [http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/Congenital-Heart-Defects_UCM_001090_SubHomePage.jsp](http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/Congenital-Heart-Defects_UCM_001090_SubHomePage.jsp)

If you want a video narrated by your high school health teacher from 1952

- [http://www.youtube.com/watch?v=5DIUk9IXUaI](http://www.youtube.com/watch?v=5DIUk9IXUaI)
Fetal Circulation

To understand how a congenital defect causes problems and why, we need to understand a little about blood flow in the fetus.
Before birth, the baby is getting all of its oxygen and nutrients from the mother through the placenta.

- Lungs are really nonfunctional at this point
  - Full of amniotic fluid, have higher resistance
  - Once the fluid is squeezed out and there is air in the lungs, the resistance to blood flow drops significantly

- Oxygen rich blood from the Mother is going to the right side of the baby’s heart, and oxygen poor blood is being pushed out to the body and back to the placenta.
Fetal Circulation

Pulmonary circuit

Pulmonary arteries → Pulmonary veins

Vena cavae

Aorta and branches

Right ventricle → Left ventricle

Systemic circuit

Arterioles

Capillary beds of all body tissues where gas exchange occurs

Venules

Oxygen-poor, CO₂-rich blood

Capillary beds of lungs where gas exchange occurs

Oxygen-rich, CO₂-poor blood

http://mcb.berkeley.edu/courses/mcb135e/difficult%20slides/Fetal%20Circulation%201.jpg
Fetal Circulation

- It is not effective to send oxygen-rich blood to the lungs at the expense of the brain.
- The baby has several 'workarounds' to allow mixing of oxygen-rich and oxygen-poor blood.
- Maximize the amount of oxygenated blood to the developing brain.
- **Ductus arteriosus**
  - Connection between pulmonary artery and aorta
- **Foramen ovale**
  - Allows blood to mix from Right Atria to Left
  - More oxygenated blood to the brain
- **Ductus venosus**
  - Fetal blood vessel connecting the umbilical vein to the IVC
  - Blood flow regulated via sphincter
  - Carries mostly high oxygenated blood
Fetal Circulation

- To summarize fetal circulation
Fetal Circulation

- After birth
  - Fluids are squeezed out of the lungs, and the resistance to blood flow drops significantly
  - Resistance to blood flow through the tissues remains about the same
    - It becomes much easier to pump blood through the lungs, and much harder to push it through the tissues
  - There is a sudden, relative pressure difference between the right and left sides of the heart
Fetal Circulation

- After birth
  - The pressure difference between the right and left heart closes the foramen ovale
    - Functionally closes at birth
  - The ductus arteriosus closes with increasing O2 concentration
    - Usually within a few days after birth
  - The ductus venosus functionally closes at birth
    - Closed within 3-7 days in term infants
Questions?

- Anything about normal cardiac anatomy or fetal development you don’t understand?
- If you don’t get something here, it’s important to ask because everything from here is built off of understanding these points.
Break
 Alright. So now that we understand a little about how the heart forms and functions, we can talk about what goes wrong.
"Congenital" is a term that means, "existing at birth"
- "Congenital Heart Defect" and "Congenital Heart Disease" tend to be used interchangeably
- The more accurate term is "defect"
- The heart or the vessels near the heart don’t develop normally before birth
The Nuts and Bolts

- Congenital Heart Defect
  - Approximately 9 in 1,000 births have a CHD
  - One of the most common birth defects
  - There is a spectrum of presentation
    - Very mild and require no treatment
    - Very severe and life threatening
  - There are currently about 1.8 million adults living with congenital heart defects
    - Likely an underestimate, because there are many mild forms that go undetected

The Nuts and Bolts

- **Congenital Heart Defect**
  - Because it can be related to signal abnormalities during development, there are other defects associated with CHD
  - VACTERL
    - Vertebral anomalies
    - Anal atresia
    - Cardiovascular abnormalities
    - Tracheoesophageal fistula
    - Esophageal atresia
    - Renal abnormalities
    - Limb defects
  - Most commonly a spontaneous genetic mutation
There is a list on the AHA website of the commonly encountered congenital heart defects. We will run through them one at a time so you better understand them. Please see the AHA website for more information. We will then group them into the common features so you have an idea what’s going on in the field.

http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/Common-Types-of-Heart-Defects_UCM_307017_Article.jsp
Congenital Heart Defects

- **Aortic Valve Stenosis**
  - The aortic valve does not properly open and close
  - Pressure builds up within the heart, and blood flow to the body is not sufficient to meet the needs of the developing baby
Congenital Heart Defects

- **Atrial Septal Defect**
  - There is a “hole” between the atria that allows mixing of blood between the two sides.
Congenital Heart Defects

- Coarctation of the Aorta
  - Narrowing of the aorta that restricts blood flow out to the body
  - Related to the genetic signaling that separates the truncus arteriosus into the pulmonary artery and the aorta
Complete AV Canal Defect

- Large hole between the atria and the ventricles that prevents separation of the oxygen rich from the oxygen poor blood
Congenital Heart Defects

- Transposition of the Great Vessels
  - The pulmonary artery and the aorta are switched
  - Right and Left sided circulation remain separate
  - Survival based on fetal communication
Ebstein’s Anomaly

- There is a malformed heart valve that does not keep blood flowing in a single direction.
- The tricuspid valve doesn’t form correctly, and allows regurgitation of blood back into the atrium.
Congenital Heart Defects

- Patent Ductus Arteriosus
  - Remnant from fetal circulation
  - Allows mixing of oxygenated blood with deoxygenated blood
  - Estimated 30% of adults have a “probable patent” PDA
Congenital Heart Defects

- **Pulmonary Valve Stenosis**
  - The pulmonic valve does not properly open and close.
  - Pressure builds up within the heart, and blood flow to the body is not sufficient to meet the needs of the developing baby.
Congenital Heart Defects

- Single Ventricle Defects
  - Rare disorder, the ventricle may be underdeveloped, small or missing a valve
  - Hypoplastic left heart
  - Pulmonary atresia
  - Tricuspid atresia
Congenital Heart Defects

- Tetralogy of Fallot
  - Four abnormalities
    - VSD, ASD, Overriding Aorta, Right Ventricular hypertrophy
  - The most common cyanotic heart disease we see
Congenital Heart Defects

- Total Anomalous Pulmonary Venous Return
  - The Pulmonary Veins do not attach to the Right Atria like they’re supposed to
  - There is mixing of the blood between the right and left heart
Congenital Heart Defects

- **Truncus Arteriosus**
  - There is one vessel carrying blood to the body and the lungs
  - Failure of the large arterial trunk to divide during embryo development
Congenital Heart Defects

- **Ventricular Septal Defect**
  - There is a “hole” between the ventricles that allows mixing of blood between the two sides
  - Very common as well
  - Large VSDs may not have a murmur at all
This seems pretty complex...

- How am I supposed to remember all of these defects and what they mean at 2am?

- Here is a somewhat simplified way of thinking of congenital heart defects
The Nuts and Bolts

- Congenital Heart Disease can best be understood by the problem it causes
  - Obstructive Defects
    - Present with low cardiac output
    - Present with shock
    - Depends on which side of the heart is obstructed
  - Left to Right shunting
    - Oxygenated blood to the deoxygenated side
    - Increased pulmonary blood flow, pulmonary congestion
    - Presentation of heart failure
  - Right to Left shunting
    - Deoxygenated blood mixing with oxygenated blood
    - Decreased pulmonary blood flow
    - Cyanosis
  - Admixture
    - Single ventricle
Obstruction

- Obstruction / Low Cardiac Output
  - Right sided obstruction
    - Pulmonary stenosis
    - Everything backs up
    - Hepatomegaly
    - Gut perfusion may be poor
      - Vomiting, poor feeding, feeding intolerance
  - Left sided obstruction
    - Pump fails pretty fast
    - Coronary blood flow is impaired
    - Developing hypoxemia
    - Weak or absent pulse
    - Cool extremities, rapidly progressing into shock
Left to Right Shunting

- Think of it as “too much blood to the lungs”
  - Poor gas exchange
- Will present looking like “Heart Failure”
- PDA, ASD, VSD, patent ductus, AV canal
- Tachypneic, increased caloric requirement
  - Small for age, poor weight gain
  - “failure to thrive”
Left to Right Shunting

- Ductus
- ASD, VSD
- AV Canals
  - Most are fairly mild
Right to Left Shunting

- Obstruction or absence of pulmonary blood flow for some reason
  - Tetralogy of Fallot, pulmonary atresia, tricuspid atresia
  - Initially do okay because of the ductus arteriosus
  - Allows some effective blood flow to get out to the lungs

- As the ductus closes during the first week or so, then they get into trouble
  - Developing hypoxemia, worsening metabolic acidosis, tachycardia

- Present with cyanosis
Right to Left Shunting

- Cyanotic Heart Disease
  - Truncus arteriosus
  - Transposition of the Great Vessels
  - Tricuspid atresia
  - Tetralogy of Fallot
  - Total Anomalous Pulmonary Venous Return

- Others
  - Ebstein’s anomaly, pulmonary atresia, hypoplastic left heart
Right to Left Shunting

- Truncus Arteriosus

http://www.riversideonline.com/health_reference/Disease-Conditions/
Right to Left Shunting

- Transposition of the Great Vessels

http://www.riversideonline.com/health_reference/Disease-Conditions/
Right to Left Shunting

- Tricuspid Atresia

[Image: Heart diagram showing Tricuspid Atresia with labels for Aorta, Patent ductus arteriosus, Pulmonary artery, Left ventricle, Right atrium, Right ventricle, Absent tricuspid valve, Atrial septal defect or Patent foramen ovale, Ventricular septal defect, Patent ductus arteriosus.]

http://www.riversideonline.com/health_reference/Disease-Conditions/
Right to Left Shunting

- Tetralogy of Fallot

- Pulmonary artery
- VSD
- Pulmonary stenosis
- Right ventricular hypertrophy
Right to Left Shunting

- Total Anomalous Pulmonary Venous Return
Double Rainbow? What Does it Mean??

- Why are we spending so much time talking about such a complicated issue?
- Good question!
Double Rainbow? What Does It Mean??

- Why are we spending so much time talking about such a complicated issue?
  - Good question!
- More than ½ of undiagnosed congenital heart defects are missed during neonatal exams
- 1/3 are missed during the 6 week exam
- Pediatrics at the UW is becoming increasingly active with the CHD kids
  - Used to all be sent to Milwaukee or Iowa City
  - There are more children in our community who are being managed for CHD
Double Rainbow? What Does It Mean??

- Why are we spending so much time talking about such a complicated issue?
  - The more mild cases may present like an upper respiratory infection
  - Even the sicker kids still have a huge differential
    - Sepsis
    - Shock
  - Infants with heart failure do not present like adults with heart failure
    - Irritable, poor feeding, failure to thrive
Take Home Points

- If you take away one slide from this presentation...
  - Think Congenital Heart Defect
  - Pink children with respiratory distress, consider one of the Left to Right shunts
  - Cyanotic children with little respiratory distress, consider one of the Right to Left shunts or a mixing lesion
  - Gray, shocky babies not responding to fluids may have an outflow tract obstruction
- All of these children need urgent evaluation in the ED!
Take Home Points

- Prehospital Treatment
  - Have a high index of suspicion for an undiagnosed Congenital Heart Defect
  - Listen closely for murmurs, especially in infants with sudden decompensation within the first month of life
  - Could be an indication that changing from fetal circulation is causing their problem
Prehospital Treatment

- Assess for perfusion and central cyanosis
  - If they are cyanotic and questionable respiratory distress
    - Think ‘not enough blood to the lungs’
      - Likely a Right to Left shunt, which is responsive to pressure
    - They need fluids, and supplemental O2
  - If they are pink and in respiratory distress
    - Think ‘too much blood to the lungs’
      - Could be a Left to Right shunt, and a ductal dependent lesion, which closes based on O2 concentration
    - Do not give big boluses as they’re already ‘wet’
    - Be judicious with O2, as it may cause their ductus arteriosus to close and make them sicker
- If they’re shocky
  - Give fluids and move quickly to the ED
Congenital Heart Defects

- **Treatment**
  - In the Emergency Department
    - If it’s a “ductal dependent lesion”, we will give them prostaglandin
    - If it’s a left to right shunt
      - Too much blood to the lungs
      - Diuresis
    - If it’s a Right to Left shunt
      - Not enough blood to the lungs
      - Give volume
  - Obstructive lesions
    - Volume and pressors
    - Prostaglandins to keep the ductus open
  - Personally, I will be calling Peds Cardiology early - and probably often!
    - Any ‘heads up’ information would be hugely appreciated
  - Most of the time, surgical correction and/or aggressive medical management is required
Congenital Heart Defects

- The good news...
  - We are getting much better at diagnosing these
    - Screening fetal ultrasounds
    - Screening ‘at risk’ patients
      - Down syndrome patients
    - A lot of the ‘bad lesions’ will hopefully be picked up early
Summary

- Congenital Heart Defects are complicated!
  - It’s amazing that anything develops correctly. EVER.
- Be sure to consider congenital heart defects in the newborn and newly-born that have sudden decompensation
- Remember the basics of evaluation and treatment
  - Blue babies that don’t have increased respiratory drive
    - May be a right-to-left shunt
  - Pink babies that look tachypneic and stressed
    - May be a left-to-right shunt
  - Gray and shocky babies
    - May have an outflow obstruction
    - May be septic!!
- There are more CHD kids in our community, coming to be seen/treated at UW
Questions?

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